Dubin-Johnson syndrome

Dubin-Johnson syndrome is a condition characterized by jaundice, which is a yellowing of the skin and whites of the eyes. In most affected people jaundice appears during adolescence or early adulthood, although a few individuals have been diagnosed soon after birth. Jaundice is typically the only symptom of Dubin-Johnson syndrome, but some people also experience weakness, mild upper abdominal pain, nausea, and/or vomiting.

Frequency

Although Dubin-Johnson syndrome occurs in people of all ethnic backgrounds, it is more common among Iranian and Moroccan Jews living in Israel. Studies suggest that this disorder affects 1 in 1,300 Iranian Jews in Israel. Additionally, several people in the Japanese population have been diagnosed with Dubin-Johnson syndrome. This condition appears to be less common in other countries.

Genetic Changes

Dubin-Johnson syndrome is caused by mutations in the *ABCC2* gene. The *ABCC2* gene provides instructions for making a protein called multidrug resistance protein 2 (MRP2). This protein acts as a pump to transport substances out of the liver, kidneys, intestine, or placenta so they can be excreted from the body. For example, MRP2 transports a substance called bilirubin out of liver cells and into bile (a digestive fluid produced by the liver). Bilirubin is produced during the breakdown of old red blood cells and has an orange-yellow tint.

ABCC2 gene mutations lead to a version of MRP2 that cannot effectively pump substances out of cells. These mutations particularly affect the transport of bilirubin into bile. As a result, bilirubin accumulates in the body, causing a condition called hyperbilirubinemia. The buildup of bilirubin in the body causes the yellowing of the skin and whites of the eyes seen in people with Dubin-Johnson syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- DJS
- hyperbilirubinemia II
- Jaundice, Chronic Idiopathic

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Dubin-Johnson syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0022350/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Bilirubin
  https://medlineplus.gov/ency/article/003479.htm
- MedlinePlus Encyclopedia: Dubin-Johnson syndrome
  https://medlineplus.gov/ency/article/000242.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Bilirubin
  https://medlineplus.gov/ency/article/003479.htm
- Encyclopedia: Dubin-Johnson syndrome
  https://medlineplus.gov/ency/article/000242.htm
- Health Topic: Jaundice
  https://medlineplus.gov/jaundice.html

Genetic and Rare Diseases Information Center

- Dubin-Johnson syndrome
  https://rarediseases.info.nih.gov/diseases/6289/dubin-johnson-syndrome

Educational Resources

- Boston Children's Hospital: Jaundice in Children
  http://www.childrenshospital.org/conditions-and-treatments/conditions/jaundice
- Disease InfoSearch: Dubin-Johnson Syndrome
  http://www.diseaseinfosearch.org/Dubin-Johnson+Syndrome/2338
- MalaCards: dubin-johnson syndrome
  http://www.malacards.org/card/dubin_johnson_syndrome
- Merck Manual Consumer Version: Jaundice in Newborns
  http://www.merckmanuals.com/home/children-s-health-issues/problems-in-newborns/jaundice-in-newborns
- Orphanet: Dubin-Johnson syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=234

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
  http://www.climb.org.uk/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/dubin-johnson-syndrome/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Jaundice%2C+Chronic+Idiopathic%22+OR+%22Dubin-Johnson+syndrome%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Dubin-Johnson+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- DUBIN-JOHNSON SYNDROME
  http://omim.org/entry/237500

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